Scientific Programme

WEDNESDAY, 31st MAY

13.30 Registration of participants

14.00 Welcome and meeting introduction: ***A. Toscano (Messina), G. Siciliano (Pisa), L. Provinciali (Ancona)***

14.30 – 15.50 WORKSHOP-1: "Distal myopathies: a clinical and therapeutic update"
Chairpersons: ***G.P. Comi, C. Fiorillo***

14.30 Emerging clinical aspects. ***E. Pegoraro***

14.45 Pathogenesis and myopathology. ***M. Mora***

15.00 Muscle imaging. ***G. Tasca***

15.15 GNE Myopathy: update and therapeutic perspectives. ***M. Mirabella***

15.30 *Discussion*

15.50 LECTURE-1
Chairperson: ***M. Filosto***

"An overview of EURO-NMD a European Reference Network for Rare Neuromuscular Diseases" ***T. Evangelista (Newcastle)***

16.30 – 16.50 *Coffee break*

16:50 – 18:50 Round Table: "Clinicians and patients associations roles: state of the art and new collaborative perspectives"

The goal of this round table is to facilitate the relationships between clinicians and patients associations as regard a better management of patients with dystrophies, myopathies, motoneuron disorders and genetic and disimmune neuropathies

Chairpersons and discussants: ***L. Politano, P. Santantonio (Mitocon), M. Marra (CIDP Onlus ITALIA), A. Toscano, D. Lauro (Famiglie SMA), G. De MARTINO (M.i.A.)***

- CAMN (Coordinamento Associazioni Malattie Neuromuscolari): a year later. ***M.L. Solinas - CAMN***
- New LEA (Livelli Essenziali di Assistenza): "pros and cons". ***M. Rasconi- UILDM***
- The age transition: an emerging problem. ***G. Siciliano***
- Data sharing and biobanks: how can we improve clinical and scientific outcomes. ***M. Moggio***
- Giornata per le Malattie Neuromuscolari (GMN): first outcomes and future proposals. ***A. Schenone***
- *Discussion*

18.50 – 20.10 WORKSHOP-2: "From clinical research into clinical practice in Duchenne Muscular Dystrophy"

Chairperson: ***G. Vita***

18.50 Introduction and welcome. ***G. Vita***

19.05 From Genetic diagnosis to personalized medicine in DMD. ***E. Pegoraro***

19.20 New standard treatment in DMD. ***M. Pane***

19.35 Observational registries: how they evolve after drugs approval. ***E. Mercuri***

19.50 *Discussion and conclusions.* ***G. Vita***

20:30 ***Welcome cocktail***

THURSDAY, 1st JUNE

07.30 – 08.30 BREAKFAST SEMINAR: "A multidisciplinary approach for the DMD patients management"

Chairperson: ***G. D'Angelo***

07.30 Guidelines for an early diagnosis and its advantages. ***S. Messina***

07.45 Management of cardiac complications. ***R. Adorisio***

08.00 Respiratory urgencies and complications. ***A. Vianello***

08.15 *Discussion*

08.30 – 09.30 Oral Communications 1- Metabolic myopathies

Chairpersons: ***S. Ravaglia, P. Tonin***

- Polymorphisms of the *GAA* gene causing amino-acid changes and analysis of the impact on protein structure in a cohort of 50 Late Onset Pompe Disease (LOPD).
  Danesino C., Ravaglia S., Scotti C., De Filippi P. and The Italian GSDII Group **(Pavia)**

- IgG anti rh-GAA assessment in an Italian cohort of patients with late-onset Pompe Disease
  Filosto M., Cotti Piccinelli S., Marchesi M., Di Muzio A., Donati M.A., Galvagni A., Lerario A., Marrosu G., Moggio M., Mongini T., Musumeci O., Pegoraro E., Piras R., Ravaglia S., Sacchini M., Sancricca C., Semplicini C., Servidei S., Siciliano G., Telese R., Tonin P., Caria F., Rota S., Padovani A., Toscano A.
  (Brescia, Chieti, Firenze, Milano, Cagliari, Torino, Messina, Padova, Pavia, Roma, Pisa, Verona)

- Registries for muscle glycogenoses: the Italian side of EUROMAC.
  Martinuzzi A., Vavla M., Musumeci O., Siciliano G., Toscano A., Bruno C., Marti Seves R.
  (Conegliano, Messina, Pisa, Genova, Barcelona)

- Report on Nationwide Italian collaborative network for Muscle Glycogen Storage Disorders
  Musumeci O., Mongini T., Angelini C., Bruno C., Moggio M., Siciliano G., Tonin P., Maggi L., Martinuzzi A., Filosto M., Servidei S., Donati A., Bembi B., Marrosu G., Di Iorio G., Ravaglia S., Pegoraro E., Bertini E., Di Muzio A., Fiumara A., Massa R., Toscano A.
  (Messina, Torino, Venezia, Genova, Milano, Pisa, Verona, Conegliano, Brescia, Roma, Firenze, Udine, Cagliari, Napoli, Pavia, Padova, Chieti, Catania)

09.30 – 10.10 LECTURE-2
Chairperson: ***A. Toscano***

"How diagnostic are serological antibodies in myopathies?"
B. Schoser (Munich, Germany)

10.10 – 10.30 *Coffee break*

10.30 – 11.30 WORKSHOP-3: "Neuromuscular junction disorders"

Chairpersons: ***G. Antonini, M. Grandis***

10.30 MUSK (Muscle Specific Kinase)-positive myasthenia. ***A. Evoli***

10.45 Thymectomy today: indications and limits of different surgical procedures. ***M. Lucchi***

11.00 Eaton-Lambert syndrome: clinical variability and treatment efficacy. ***C. Rodolico***

11:15 *Discussion*

11.30 – 13.00 Oral Communications 2 – SMA/Myopathies

Chairpersons: ***L. Bello, L. Politano***

- Correlation between IGHMBP2 protein levels in human motor neuron and non-neuronal somatic cells and phenotype of 5 patients affected with spinal muscular atrophy with respiratory distress type 1
  Govoni A., Magri F., Salani S., Del Bo R., Taiana M., Forotti G., Bresolin N., Comi G.1, Nizzardo M., Corti S. **(Milano)**

- POPDC1 Gene mutations screening in Laminopathies: possible role as a modifier
  Rossi R., Scotton C., Maggi L., D'Amico A., Ricci G., Vercelli L., Benedetti S., Bertini E., Mercuri E., Rodolico C., Mongini T., Carboni N., Marrosu G., Chico L., Santoro L., Merlini L., Lattanzi G., Morandi L., Barton P.J.R., Buchan R., Walsh R., Cook S.A., Milting H., Bonne G., Brand T., Ferlini A. **(Ferrara, London, Bad Oeynhausen, Paris)**

- Glycosilation of Alpha-Dystroglycan: one pathway, several phenotypes
  Astrea G., D'Amico A., Battini R., Berardinelli A., Bertini E., Bruno C., Cassandrini D., Catteruccia M., Comi G.P., Fattori F., Fiorillo C., Giannotta M., Gorni K., Magri F., Mercuri E., Messina S., Mongini T., Mora M., Morani F., Moro F., Pane M., Pegoraro E., Pini A., Politano L., Ricci F., Sframeli M., Santorelli F.M.
  (Pisa, Roma, Pavia, Genova, Milano, Bologna, Messina, Torino)

- Next-generation sequencing approach for the diagnosis of congenital myopathies: a 3-yr experience.
  Cassandrini D., Trovato R., Rubegni A., Lenzi S., Baldacci J., Fiorillo C., Savarese M., Nigro V., Bruno C., Astrea G., Santorelli F., and Italian Network on Congenital Myopathies
  (Pisa, Genova, Napoli)

- Autosomal recessive myopathy associated with cataracts caused by mutations in the gene encoding INPP5K, an inositol phosphatase
  ***Roos A., Wiessner M., Cox D., Hathazi D., Marini-Bettolo C., Straub V., Barresi R., Senderek J., Lochmüller H*. (Newcastle upon Tyne, Dortmund, Munich)**

- Clinical expression of facio-scapulo-humeral muscular dystrophy in carriers of 33-35 kb D4Z4 reduced alleles: experience of the Italian National Registry for FSHD.
  Ruggiero L., Mele F., Ricci G., Vercelli L., Govi M., Nikolic A., Louise M., Sera F., Berardinelli A., Angelini C., Antonini G., Bucci E., Filosto M., Cao M., Giardina E., Pegoraro E., Di Muzio A., D'Amico M.C., Maggi L., Portaro S., Rodolico C., Villa L., Mongini T., Siciliano G., Tomelleri G., D'Angelo G., Maioli M.A., Moggio M., Santoro L., Tupler R.
  (Napoli, Pisa, Modena, Torino, Worcester, London, Pavia, Venezia, Roma, Brescia, Padova, Milano, Messina, Verona, Bosisio Parini, Cagliari)

13.00 – 14.00 *Lunch*

14.00 – 15.30 Poster Session 1

Chairpersons: ***M. Garibaldi, R. Piras, G. Primiano, M. Scarlato, M. Scarpelli***

P1-1 MYASTHENIA
P1-2 CONGENITAL MYOPATHIES
P1-3 INFLAMMATORY MYOPATHIES
P1-4 METABOLIC MYOPATHIES
P1-5 MITOCHONDRIAL MYOPATHIES
P1-6 OTHER MYOPATHIES

15.30 – 16.30 WORKSHOP-4: "Recent molecular diagnostic techniques: shall we get earlier diagnosis?"

Chairpersons: ***G. Ricci, S. Servidei***

15:30 Limb-Girdle Muscular Dystrophies. ***V. Nigro***

15.45 Mitochondrial disorders. ***M. Zeviani***

16.00 Facio-Scapulo-Humeral Dystrophy. ***R. Tupler***

16.15 *Discussion*

16.30 – 17.30 Oral communications 3 – Duchenne muscular dystrophy

Chairpersons: ***A. Ardissone, G. Di Iorio***

- Observational study on the nutritional and metabolic features in Duchenne muscular dystrophy: the Italian "N&M_Duchenne study"
  Bertoli S., Baranello G., Foppiani A., Giaquinto E., De Amicis R., Leone A., Battezzati A. **(Milano, Cesena)**

- Respiratory function in ataluren-treated, non-ambulatory patients with nonsense mutation Duchenne (NMDMD) muscular dystrophy from a long-term extension trial versus untreated patients from a natural history study
  Comi G.P., Bertini E., Magri F., Luo X., McIntosh J., Ong T., Riebling P., Triffilis P., Souza M., Peltz S.W., Mercuri E. **(Milano, Roma, South Plainfield)**

- Vision DMD: a drug development program for Vamorolone in Duchenne muscular dystrophy
  Guglieri, M., Clemens P., Cnaan A., Damsker J., Gordish-Dressman H., Morgenroth, L., Davis R., Smith A., Storch K., Head R., Demotes-Mainard J., Nagaraju K., Hathout Y., Athanasiou D., Vroom E., Bushby K., Hoffman E. **(New Castle upon Tyne, Pittsburgh, Washington)**

- Long-Term treatment with Eteplirsen in non-ambulatory patients: a case study in identical twins
  Lowes L.P., Alfano L., Dracker R.A., Duda P., Mendell J.R. **(Columbus, Liverpool, Cambridge)**

Social programme

a) Guided walking tour of Ortigia

b) Evening at the Greek Theater for Classical Plays (Greek Tragedies)

FRIDAY, 2nd JUNE

07.30 – 08.30 BREAKFAST SEMINAR: "ERT (Enzyme Replacement Therapy) in Pompe disease: 10 years of experience looking at the future"
Chairperson: ***C. Angelini***

07.30 A clinical and laboratory update. ***A. Toscano***

07.45 Classic and non-classic forms: treatment and management. ***A. Fiumara***

08.00 Therapy in Late 0nset forms (LOPD - Late Onset Pompe Disease): clinical assessment and future views. ***M. Moggio***

08.15 *Discussion*

08.30 – 09.30 Muscle Club: discussion of peculiar diagnostic pathways

Chairpersons: ***L. Maggi, E. Pennisi***

- Expanding clinical and histological spectrum of DNM2 mutations ***Barcellona C., Musumeci O., Savarese M., Nigro V., Toscano A., Rodolico C.*** **(Messina, Napoli)**

- Novel compound heterozygous mutations of AGRN resulting in a complex muscular phenotype ***Gemelli C., Cassandrini D., Fabbri S., Lamp M., Santorelli F.M., Bruno C., Reni L., Broda P., Fiorillo C., Mandich P., Grandis M.*** **(Genova, Pisa)**

- Myotonia in Filamin-C-related myopathies ***Lerario A., Peverelli L., Villa L., Lucchiari S., Tironi R., Conca E., Fagiolari G., Grimoldi N., Sciacco M., Comi G.P., Moggio M.*** **(Milano)**

- Core myopathy with early respiratory failure and titin gene mutation ***Petrucci A., Costanzi-Porrini S., Giacanelli M., Lispi L., Santorelli F.M., Cassandrini D., Rastelli E., Nigro V., Massa R., Savarese M.*** **(Roma, Pisa, Napoli)**

- Muscle pathological features of a hyperkalemic paralysis/dermatomyositis "double trouble" ***Rota S., Galvagni A., Caria F., Marchesi M., Cotti Piccinelli S., Baronchelli C., Padovani A., Filosto M.*** **(Brescia)**

- Castleman disease and inflammatory myopathy: is there a link? ***Simoncini C., Montano V., Ricci G., Calì L., Giorgetti M., Alì G., Rousseau M., Tavoni A., Siciliano G.*** **(Pisa)**

09.30 - 10.10 LECTURE-3
Chairperson: *R. Liguori*

"How still relevant is in muscle disorders the neurophysiological support for the diagnosis?" ***P. Girlanda (Messina)***

10.10 – 10.30 *Coffee break*

10.30 – 11.30 WORKSHOP-5: "Spinal muscular atrophy: new clinical and therapeutic aspects"

Chairpersons: ***A. Berardinelli, G. Marrosu***

10.30 Emergent phenotype and new causative genes. ***E. Bertini***

10.45 EAP (Expanded Access Program) Nusinersen experience. ***S. Messina***

11.00 A clinical trial overview. ***E. Mercuri***

11.15 *Discussion*

11.30 – 13.00 Oral Communications 4 – New results from large populations studies

Chairpersons: ***G.R. Barresi, A. Di Muzio***

- Myositis-specific antibodies (MSA): high prevalence in biopsy-proven myositis population
  Lucchini M., De Fino C., Tasca G., Primiano G., Ricci E., Silvestri G., Servidei S., Mirabella M. **(Roma)**

- NGS target re-sequencing analysis in patient with persistent asymptomatic or mildly symptomatic hyperCKemia.
  Fiorillo C., Madia F., Robbiano A., Pozzolini G., Trucco F., Pedemonte M., Diana M.C., Grandis M., Gemelli C., Fabbri S., Schenone A., Nobili F.M., Foiadelli T., Trabatti C., Savasta S., Schiaffino M.C., Picco P., Morcaldi G., Celle M.E., Mancuso M., Tonin P., Mandich P., Bruno C., Zara F.
  (Genova, Pavia, Pisa, Verona)

- NGS in neuromuscular disorders: an update
  Gibertini S., Saredi S., Pasanisi M.B., Ardissone A., Moroni I., Baranello G., Peverelli L., Tonin P., Previtali S.C., Grandis M., Gemelli C., Canioni E., Bernasconi P., Mantegazza R., Morandi L., Maggi L., Mora M., Ruggieri A. **(Milano, Verona, Genova)**

- Comprehensive CNVS assessment in 234 diagnosis-resistant myopathic patients.
  Giugliano T., Garofalo A., Savarese M., Torella A., Musumeci O., Maggi L., Ruggiero L., Vercelli L., D'Amico A., Fiorillo C., Magri F., Piluso G., De Bleecker J., Toscano A., Mora M., Santoro L., Mongini T., Bertini E., Bruno C., Comi G.P., Santorelli F.M., Angelini C., Politano L., Nigro V.
  (Napoli, Pozzuoli, Helsinki, Messina, Milano, Torino, Roma, Genova, Pisa, Padova, Belgium)

- Phenotypical and molecular definition of a multi-center cohort of 49 patients with limb girdle muscular dystrophy Type 2A
  Bello L., ***Magri F., Lazzarotto A., Semplicini C., Fanin M., Gandossini S., Diella E., D'Angelo M.G., Comi G.P., Pegoraro E.*** **(Padova, Milano, Bosisio Parini)**

- Big Data in genetic research: the example of titin gene and titinopathies
  Savarese M., Johari M., Johnson K., Torella A., Topf A., Evilä A., Arumilli M., Rubegni A., Kuhn M., Fattori F., Santorelli F.M., Schoser B., Straub V., Nigro V., Hackman P., Udd B.
  (Helsinki, Napoli, Pozzuoli, New Castle upon Tyne, Pisa, Neu-Ulm, Roma, Munich, Tampere)

13.00 – 14.00 *Lunch*

14.00 – 15.30 Posters Session 2

Chairpersons: ***G. Baranello, F. Biasini, A. D'Amico, J.F. Desaphy, F. Santorelli***

P2-1 DYSTROPHIC AND NON-DYSTROPHIC MYOTONIAS
P2-2 DYSTROPHINOPATHY/LGMD/CONGENITAL MUSCULAR DYSTROPHIES
P2-3 SMA
P2-4 MISCELLANEOUS

15.30 – 16.10 LECTURE- 4
Chairperson: ***C. Bruno***

"Early onset metabolic myopathies: from diagnosis to therapy**"** ***A. Donati (Firenze)***

16.10 – 16.30 *Coffee break*

16.30 – 18.15 Oral communications 5 - Myopathies/Dystrophic and non-dystrophic myotonias

Chairpersons: ***R. Massa, L. Ruggiero***

- The importance of muscle biopsy for muscle diseases experts: a review of Sardinian data
  Piras R., ***Maioli M.A., Murru M.R., Costa G., Solla E., Mancosu C., Mammoliti R., Marrosu G.*** **(Cagliari)**

- Very late-onset non-thymomatous and thymomatous myasthenia gravis (MG) are associated with different HLA class II alleles
  Massa R., ***Greco G., Testi M., Antonini G., Marfia G.A., Rastelli E., Terracciano C., Pompeo E., Andreani M.*** **(Roma)**

- 20 years clinical follow-up in patients with oculopharyngeal muscle disease (OPMD)
  Brizzi T., ***Barca E., Biasini F., Lupica A., Vita G., Mazzei R.L., Musumeci O., Rodolico C., Toscano A.*** **(Messina, Palermo, Cosenza)**

- Assessing the impact of gender on the phenotype of Myotonic dystrophy type 2: a cohort of 307 patients
  Montagnese F., ***Mondello S., Wenninger S., Schoser B.*** **(Munich, Messina)**

- A respiratory snapshot of a cohort of DM1 patients: clinically stable does not mean normal.
  Falcier E., ***De Mattia E., Sannicolò G., Lizio A., Iatomasi M., Lupone S., Mollar E., Castino V., Roma E., Conti C., Rao F., Sansone V.A. (Milano)***

- Precision medicine to address therapy in myotonia caused by sodium channel mutations
  Farinato A., ***Altamura C., Imbrici P., Maggi L., Mantegazza R., Filosto M., Siciliano G., Sansone V.A.,***
  Lo Monaco M., the Italian Network for Muscle Channelopathies, Conte Camerino D., Desaphy J-F.
  (Bari, Milano, Brescia, Pisa, Roma)

- Novel zebrafish models of sarcoglycanopathy
  Soardi M., ***Carotti M., Fecchio C., Sacchetto R., Sandonà D.*** **(Padova)**

18.15 – 19.45 Annual meeting of AIM members

20.45 *Social dinner*

SATURDAY, 3rd JUNE

08.30 – 09.30 Discussion on AIM projects in progress

Chairpersons: ***A. Martinuzzi, G. Siciliano***

- The nation-wide Italian collaborative network of mitochondrial diseases: from 2009 to today
  Mancuso M., Caldarazzo Ienco E., Angelini C., Bertini E., Pegoraro E., Carelli V., Comi G.P., Minetti C., Bruno C., Moggio M., Mongini T.,Vercelli L., Primiano G., Servidei S., Orsucci D., Tonin P., Toscano A., Musumeci O., Moroni I., Santorelli F.M., Filosto M., Lamperti C., Zeviani M., Siciliano G.
  (Pisa, Venezia, Roma, Bologna, Milano, Genova, Torino, Verona, Messina, Brescia)

- TREAT-CDM, an international observational longitudinal study on congenital myotonic dystrophy (CDM): a spin-off for the creation of the Italian CDM network
  Sansone V., Albamonte E., Casiraghi J., Pinia A., Berardinelli A., D'Angelo G., D'Amico A., Bertini E., Ricci F., Ardissone A., Filippini M., Conti C., Moscardi M., Morettini V., Iatomasi M., De Biaggi M.L., Maestri E., Zanolini A., Pane M., Battini R., Astrea G., Baranello G., Moroni I., Messina S., Mercuri E., Bruno C., Fiorillo C., Siciliano G., Baldanzi S., Fossati B., Mongini T., Vita G., Meola G., Rodolico C., Toscano A., Campbell C., Johnson N.
  (Milano, Bologna, Pavia, Bosisio parini, Roma, Torino, Pisa, Messina, Genova, Ontario, Utah)

- Muscle pain in mitochondrial diseases: a survey on a large cohort from the Italian Network
  Filosto M., Cotti Piccinelli S., Marchesi M., Angelini C., Bertini E., Bruno C., Caldarazzo Ienco E., Carelli V., Comi G.P., Lamperti C., Minetti C., Moggio M., Mongini T., Moroni E., Musumeci O., Orsucci D., Pegoraro E., Primiano G., Santorelli F.M., Servidei S., Simoncini C., Tonin P., Toscano A., Vercelli L., Zeviani M., Galvagni A., Caria F., Rota S., Padovani A., Siciliano G., Mancuso M.
  (Brescia, Venezia, Roma, Genova, Pisa, Bologna, Milano, Torino, Messina, Padova, Verona, Cambridge)

- CARE-DMD, A LONGITUDINAL STUDY ON DMD CARE: Energy expenditure and muldisciplinary involvement at transitional age and towards adult life in DMD
  Messina S., Berardinelli A., D'Angelo G., D'Amico A., Pane M., Bruno C., Sansone V.A., Mongini T., Pegoraro E., Comi G.P., Moggio M., Previtali S., Mercuri E., Vita G.
  (Messina, Bosisio Parini, Roma, Genova, Milano, Torino, Padova)

09.30 – 10.10 Lecture-5

Chairperson: ***O. Musumeci***

"Therapy and diagnostic functional testing in metabolic myopathies"
J. Vissing (Copenhagen, Denmark)

10.10 – 10.30 *Coffee break*

10.30 – 11.15 WORKSHOP-6: "Recent therapeutic approaches in Muscle disorders"

Chairpersons: ***C. Minetti, V. Sansone***

10.30 Innovative treatments in Muscular Dystrophies. ***T. Mongini***

10.45 Experimental approach in Merosin-Deficient Congenital Muscular Dystrophy. ***S. Previtali***

11.00 Therapeutic perspectives in dystrophic and non-dystrophic myotonias. ***G. Meola***

11.15 Mitochondrial medicine: what`s new?. ***M. Mancuso***

11.30 *Discussion*

11.45 – 13.30 Oral Communications 6 – Clinical and experimental DMD features

Chairpersons: ***A. Ferlini, M. Sciacco***

- The mildest end of the dystroglycanopathy phenotypic spectrum: from asymptomatic hyperCKemia to limb girdle muscular dystrophy
  Brisca G., Pedemonte M., Trucco F., Ferretti M., Diana M.C., Magnano G.M., Valle M., Broda P., Minetti C., Fiorillo C., Bruno C. **(Genova)**

- Tele-monitoring in paediatric ventilated neuromuscular patients: results of an Italian multicentric study
  Trucco F., Pedemonte M., Romano C., Wenzel A., Tacchetti P., Bella C., Falsaperla R., Racca F., Bruno C., Minetti C. **(Genova, Alessandria, Catania)**

- Insights into bone mineral density and bone metabolism in Duchenne muscular dystrophy.
  Vita GL., Catalano A., Sframeli M., Distefano MG., La Rosa M., Barcellona C., Bonanno C., Nicocia G., Profazio C., Morabito N., Lunetta C., Vita G., Messina S. **(Messina)**

- Development of a human cardiac organoid to study heart dysfunctions in Duchenne and Becker muscular dystrophies
  Piga D., Rinchetti P., Salani S., Calandriello L., Forotti G., Nizzardo M., Magri F., Comi G.P., Corti S. **(Milano)**

- Multilevel molecular analysis identifies all dystrophin gene mutations pointing out that DMD is a genetically homogenous disease: repercussions on diagnosis, prevention and therapy
  Neri M., Selvatici R., Falzarano M.S., Trabanelli C., Ravani A., Rimessi P., Fabris M., Scotton C., Mauro A., Fortunato F., Osman H, Rossi R., Armaroli A., Buldrini B., Merlini L., Mora M., Gorni K., Sansone V., Mongini T., Pegoraro E., Tonin P., Fiorillo C., Pini A., Filosto M., D'Amico A., Bertini E., Messina S., Vita G., Pane M., Mercuri E., Muntoni F., Fini S., Gualandi F., Ferlini A.
  (Ferrara, Milano, Torino, Padova, Verona, Genova, Bologna, Brescia, Roma, Messina, London)

- Fibroblasts-derived exosomes: potential role in the fibrotic process of Duchenne muscle dystrophy
  Zanotti S., Gibertini S., Blasevich F., Saredi S., Maggi L., Bragato C., Ruggieri A., Mantegazza R., Mora M. **(Milano)**

- Chromatin configuration RNA and protein studies identified novel DNA elements that influence the dystrophin transcription dynamics
  Falzarano M.S., Gherardi S., Bovolenta M., Passarelli C., Erriquez D., Scotton C., Armaroli A., Rossi R., Osman H., Mora M., Bernasconi P., Maggi L., Morandi L., Perini G., Ferlini A.
  (Ferrara, Bologna, Roma, Milano, London)

13.30 – 13.45 Closing remarks

13.45 – 14.00 ECM Questionnaire

POSTER SESSION – 1st June 2017, 14.00 – 15.30

Chairpersons: ***M. Garibaldi, R. Piras, G. Primiano, M. Scarlato, M. Scarpelli***

P1-1 MYASTHENIA

P.1 Thymoma-associated Myasthenia Gravis: clinical and serological features of Pisa's Cohort
De Rosa A., Ricciardi R., Maestri M., Guida M., Rizzo S., Lucchi M., Mussi A., Bonuccelli U. **(Pisa)**

P.2 Clinical, morphological and immunological findings in myasthenia – myositis association
Garibaldi M., Fionda L., Bucci E., Vizzaccaro E., Di Pasquale A., Morino S., Pennisi E.M., Antonini G. **(Roma)**

P.3 A case with chronic inflammatory demyelinating polineuropathy and ocular myasthenia gravis
Montano V., Maestri M., Ricci G., Alì G., Mancuso M., Ricciardi R., Siciliano G. **(Pisa)**

P.4 Impact of myasthenia gravis on quality of life
Lupidi F., Carlini G., Provinciali L., Logullo F. **(Ancona, Macerata)**

P1-2 CONGENITAL MYOPATHIES

P.5 A Novel ACTA1 mutation in a patient affected by congenital myopathy with histopathologic progression
Cuccagna C., Fattori F., Primiano G., Sancricca C., Bernardo D., Sauchelli D., Verardo M., Bertini E., Servidei S. **(Roma)**

P.6 Pseudo-dominant inheritance of a novel homozygous HACD1 mutation associated with congenital myopathy: the first Caucasian family
Emmanuele V., Savarese M., Musumeci O., Brizzi T., Torella A., Conca E., Nigro V., Moggio M., Toscano A., Rodolico C. **(Messina, Pozzuoli, Milano)**

P.7 Fetal akinesia deformation sequence and recessive central core disease: a rare presentation of mutations in RYR1 gene
Emmanuele V., Torella A., Sframeli M., Musumeci O., Messina S., Nigro V., Rodolico C., Toscano A. **(Messina, Pozzuoli)**

P.8 Congenital myopathy with protein aggregates and nemaline bodies related to CFL2 mutations
Fattori F., Fiorillo C., Rodolico C., Tasca G., Verardo M., Bellacchio E., Fagiolari G., Lupica A., Broda P., Moggio M., Bruno C., Tartaglia M., Bertini E., D'Amico A. **(Roma, Genova, Messina, Milano)**

P.9 Neuropsicological pattern in centronuclear myopathy due to DNM2 gene mutations
La Rosa C., Sframeli M., Vita G.L., Di Stefano M.G., La Rosa M., La Foresta S., Faraone C., Russo M., Fattori F., D'Amico A., Bertini E., Rodolico C., Lunetta C., Messina S., Vita G. **(Messina, Pisa, Roma)**

P1-3 INFLAMMATORY MYOPATHIES

P.10 Atypical clinical pictures in inflammatory myopathies: a case series
Caria F., Galvagni A., Baronchelli C., Rota S., Gallo Cassarino S., Marchesi M., Cotti Piccinelli S., Padovani A., Filosto M. **(Brescia)**

P.11 Statin-induced necrotizing autoimmune myopathy: clinical, hystopathological and radiological characterization of five patients
Villa L., Peverelli L., Lerario A., Tironi R., Ciscato P., Woods A.M., Sciacco M., Moggio M. **(Milano, Oxford)**

P.12 Muscle biopsy findings and outcome in necrotizing autoimmune myopathy
Amati A., Lia A., Girolamo F., Serlenga L., Giannini M., Iannone F., Trojano M. **(Bari)**

P1-4 METABOLIC MYOPATHIES

P.13 Asyntomatic primary carnitine deficiency unmasked in a mother by newborn screening
Cotti Piccinelli S., Marchesi M., Carducci C., Angeloni A., Rota S., Caria F., Galvagni A., Baronchelli C., Padovani A., Filosto M. **(Brescia, Roma)**

P.14 Atypical features in multiple ACYL-COA dehydrogenase deficiency: report of two cases
Lupica A., Musumeci O., Barca E., Mazzeo A., Rodolico C., Toscano A. **(Messina)**

P.15 Dilative Arterial malformations in patients with Late Onset Pompe Disease (LOPD)
Musumeci O., Granata F., Rodolico C., Arrigo R., Mosca V., Brizzi T., Ciranni A., Longo M., Toscano A. **(Messina)**

P.16 Riboflavin transporter deficiency (BVVL): transient expanded newborn screening (NBS) positivity for beta-oxidation abnormalities
Pasquini E., Sacchini M., Cavicchi C., Malvagia S., Funghini S., Donati M.A. **(Firenze)**

P.17 Newborn screening for Pompe disease in Tuscany and Umbria: current overview and first preliminary results after two years
Pasquini E., La Marca G., Morrone A., Daniotti M., Forni G., Catarzi S., Scolamiero M., Sacchini M., Donati M.A. **(Firenze)**

P.18 Development of a mobile app conceptually designed for patients with Pompe disease
Peviani S., Carlini F., Proietti C., Ricci G., Vianello A., Baldanzi S., Ubaldi U., Seidita F., Antonini G., Siciliano G. **(Pisa, Milano, Padova, Roma)**

P.19 Polymorphisms in exercise genes and respiratory outcome after ERT in a cohort of Late Onset Pompe Disease (LOPD)
Ravaglia S., Carlucci A., Malovini A., Danesino C., De Filippi P. and the Italian GSDII Group **(Pavia)**

P.20 Functional assessment tools in infantile Pompe disease. A critical analysis and pilot study
Ricci F., Brusa C., Berardinelli A., Rolle E., Rossi F., Placentino V., Spada M., Pagliardini V., Vitiello B., Mongini T. **(Torino, Pavia)**

P.21 Three Dimensional Gait Analysis in Late Onset Pompe Disease (LOPD)
Sancricca C., Rossellini G., Denza G., Pelliccioni M., Primiano G., Cuccagna C., Bernardo D., Sauchelli D., Servidei S. **(Roma)**

P.22 Young girl complaining of fatigue and muscle contractures
Agazzi E., Rottoli M.R. **(Bergamo)**

P.23 The importance of a non-invasive screening in proximal myopathies
Sampaolo S., Allegorico L., Bruno G., Lombardi L., Di Iorio G. **(Napoli)**

P1-5 MITOCHONDRIAL MYOPATHIES

P.24 Familial ALS, clinical heterogeneity and mitochondrial disorders: description of a family
Bisordi C., LoGerfo A., Caldarazzo Ienco E., Mancuso M., Siciliano G. **(Pisa)**

P.25 Mitochondrial Involvement in Patients with Autism spectrum disorders
Borgione E., Lo Giudice M., Castello F., Santa Paola S., Musumeci S.A., Giusto S., Di Vita G., Vitello G.A., Di Blasi F., Savio M., Scuderi C. **(Troina)**

P.26 Mitochondrial Giant Crystals in muscle biopsy
Costa R., Papa V., D'Angelo R., Rinaldi R., Tonon C., Lodi R., Cenacchi G. **(Bologna)**

P.27 Liver transplantion reverses biochemical imbalance and improves clinical conditions in mitochondrial neurogastrointestinal encephalomyopathy
Rinaldi R., D'Angelo R., Pironi L., Pinna A.D., Caporali L., Boschetti E., Cenacchi G., Lodi R., Carelli V., De Giorgio R. **(Bologna)**

P.28 Growth Differentiation Factor 15 as a useful biomarker for miochondrial Disorder
Salvatore S., Formichi P., Taglia I., Bracalente I., Battisti C., Malandrini A., Federico A. **(Siena)**

P1-6 OTHER MYOPATHIES

P.29 Discordant manifestations in two Italian brothers with GNE myopathy
Dotti M.T., Lornage X., Malandrini A., Bohm J., Romero N.B., Laporte J., Malfatti E. **(Siena, Illrich, Paris)**

P.30 A PGM NGS protocol in a single center cohort for patients with undiagnosed myopathy
Marchesi M., Lanzi G., Galvagni A., Cotti Piccinelli S., Mori L., Caria F., Rota S., Gallo Cassarino S., Facchetti F., Padovani A., Giliani S., Filosto M. **(Brescia)**

P.31 Severe muscle involvement caused by A193T mutation in FILAMIN-C
Monforte M., Ricci E., Udd B., Tasca G. **(Roma, Helsinki, Tampere, Vaasa)**

P.32 Clinical next generation sequencing gene Panel in patients orphan of genetic diagnosis
Neri M., Selvatici R., Scotton C., Storbeck M., Vezyroglou K., Heller R., Tugnoli V., Bigoni S., Timmerman V., Wirth B., De Grandis D., Gualandi F., Ferlini A. **(Ferrara, Cologne, Antwerpen, Verona)**

P.33 The success of whole exome sequencing analysis in neuromuscular diseases patients: the unife experience within neuromics project
Neri M., Scotton C., Selvatici R., Gualandi F., Wirth B., Schols L., Klockgether T., Lochmuller H., Muntoni F., D'Amico A., Bertini E., Pane M., Mercuri E., Ferlini A.
(Ferrara, Cologne, Tübingen, Bonni, Newcastle upon Tyne, London, Roma)

P.34 Systemic al amyloidosis revealed by a muscle biopsy: a case report
Papa V., Costa R., Bellanova M.F., Capozzi A.R., Saccani E., Verga L., Capello G.L., Accardi F., Giuliani N., Pietrini V., Cenacchi G. **(Bologna, Parma, Pavia)**

P.35 GNE myopathy functional activity scale (GNEM-FAS): a four year follow up in 10 HIBM patients
Parisi D., Portaro S., Brizzi T., Biasini F., Cavallaro F., Vita G., Toscano A., Rodolico C. **(Messina, Palermo)**

P.36 McLeod syndrome: an Italian familiy with a novel mutation in the XK gene.
Piccolo G., Tartara E., Terzaghi M., Cortese A., Cittadella R., Benna P., Cavallaro S., Galimberti C.A.
(Pavia, Catania, Torino)

P.37 Detection and multidisciplinary care of myopathic patients in Ogliastra
Piras R., Maioli M.A., Murru M.R., Costa G., Solla E., Mancosu C., Mammoliti R., Marrosu G. **(Cagliari)**

P.38 PLEC gene mutations cause familial disto-proximal myopathy and long QT syndrome mimicking mitochondrial disease
Primiano G., Tartaglia M., Cuccagna C., Sauchelli D., Bernardo D., Sancricca C., Lucchini M., Mirabella M., Servidei S. **(Roma)**

P.39 Myalgias, cramps and muscle rippling: a case report
Ricci G., Alì G., Chico L., Logerfo A., Cassandrini D., Comi G., Sorrentino V., Costa R., Cenacchi G., Siciliano G. **(Pisa, Milano, Siena, Bologna)**

P.40 Encephalocardiomyopathy with severe recurrent rhabdomyolysis due to TANGO2 mutations: a case report
Ricci F., Brusa C., Ferraris S., Ferrero G.B., Bertini E., Di Rocco M., Moroni I., Larosa P., Zoppo M., Manole A., Scalco R., Vitiello B., Mongini T. **(Torino, Roma, Genova, Milano, London)**

P.41 Next-generation sequencing approach for the diagnosis of genetic basis of hyperckemia: results from 25 patients
Rubegni A., Astrea G., Battisti C., Cassandrini D., Donati M.A., Dotti M.T., Lenzi S., Maioli M.A., Malandrini A., Melani F., Pasquini E., Trovato R., Santorelli F.M. **(Pisa, Siena, Firenze, Cagliari)**

P.42 Expression of Aquaporin 4 in normal human muscle is independent from myosin heavy chain isoform
Vizzaccaro E., Terracciano C., Rastelli E., Massa R. **(Roma)**

P.43 Benign monomelic amyotrophy of lower limb: report of 15 Italian cases
Di Muzio A., Barbone F., Telese R. **(Chieti)**

P.44 Quantitative muscle ultrasound analysis in neuromuscular disorders
Di Pasquale A., Fragiotta G., Vanoli F., De Santis T., Fionda L., Garibaldi M., Bucci E., Vizzaccaro E., Morino S., Antonini G. **(Roma)**

P.45 Neurorehabilitation in ALS: consequences at micrornas level
Giaretta L., Pegoraro V., Merico A., Angelini C. **(Venezia)**

P.46 The Questionnaire GNAMM: the eating habits of 436 people with neuromuscular disease in Italy.
Pini A., Zoni L., Poli C., Giannotta M., Di Pisa V., Siciliano G., Santorelli F., Mongini T., Filosto M., Politano L. **(Bologna, Pisa Torino, Brescia, Napoli)**

P.47 BAG3 mutation: from cardiomyopathy to a complex severe neuromuscular disorder with myofibrillar myopathy in a pediatric case
Scarpini G., Di Pisa V., Giannotta M., Nigro V., Ragni L., Valentino L., Pini A. **(Bologna, Napoli)**

P.48 A singular case of Rhabdomyolisis with reversible paralysis
Rastelli E., Vizzaccaro E., Frezza E., Massa R. **(Roma)**

POSTER SESSION – 2nd June 2017, 14.00 – 15.30

Chairpersons: ***G. Baranello, F. Biasini, A. D'Amico, J.F. Desaphy, F. Santorelli***

P2-1 DYSTROPHIC AND NON-DYSTROPHIC MYOTONIAS

P.49 Functional study of myotonia congenita mutations in the C-terminus of CLC-1 and proof of concept study for chaperone-mediated rescue of trafficking-defective CLC-1 mutant
Altamura C., Imbrici P., Lucchiari S., Sahbani D., Comi G.P., Dotti M.T., Meola G., Politano L., Lo Monaco M., Desaphy J.F., Conte Camerino D.
(Bari, Milano, Siena, Napoli)

P.50 Myotonic dystrophy type 2 in a Sicilian cohort: a challenging diagnosis by biomolecular tests
Biasini F., Rodolico C., Brizzi T., Cardani R., Valaperta R., Toscano A., Meola G., **(Messina, Milano)**

P.51 Cardiac troponin T in skeletal muscle from myotonic dystrophies patients: a possible biomarker of cardiac dysfunctions
Bose' F., Renna L.V., Ferrari N., Arpa G., Fossati B., Meola G., Cardani R. **(Milano)**

P.52 Skeletal muscle CLC-1 channel: from gene to protein, from birth to aging
Conte E., Fonzino A., Cibelli A., De Luca A., Pierno S., Nicchia G.P., Camerino G.M. **(Bari)**

P.53 Mild phenotype in DM1 young boy due to interrupted repeat of the DMPK expanded tract
Fossati B., Cardani R., Valaperta R., Cavalli M., Brigonzi E., Meola G. **(Milano)**

P.54 Monitoring motor function and disease progression in DM1
Gualandris M., Maestri E., Lizio A., Gatti V., Paci E., Beshiri F., Zanolini A., Sansone V. **(Milano)**

P.55 Gender-related characteristics of myotonic dystrophy type 1 in a large Italian database
Lozzi F., Rastelli E., Antonini G., Bianchi M.L.E., Botta A., Bucci E., Casali C., Costanzi Porrini S., Frezza E., Giacanelli M., Greco G., Inghilleri M., Novelli G., Pennisi E.M., Petrucci A., Piantadosi C., Silvestri G., Terracciano C., Vanacore N., Massa R. **(Roma)**

P.56 Non dystrophic myotonias: review of our cases with focus on genotype-phenotype correlations and therapeutic effects of Mexiletine
Montano V., Ricci G., Simoncini C., Chico L., Bernasconi P., Lehmann-Horn F., Siciliano G.
(Pisa, Milano, Ulm)

P.57 Thomsen disease with central core features at muscle biopsy; a new morfhological pattern or an unusual double trouble?
Peverelli L., Villa L., Lerario A., Lucchiari S., Pagliarani S., Tironi R., Conca E., Fagiolari G., Grimoldi N., Sciacco M., Comi G.P., Moggio M. **(Milano)**

P.58 Post-receptor abnormalities contribute to insulin resistance in myotonic dystrophy type 1 and type 2 skeletal muscle
Renna L.V., Iachettini S., Fossati B., Saraceno L., Colombo R., Meola G., Cardani R. **(Milano)**

P.59 Non invasive ventilation in DM1: evaluation of compliance in a cohort of patients followed at the Nemo Center Milan
Sannicolò G., De Mattia E., Falcier E., Lizio A., Iatomasi M., Lupone S., Mollar E., Castino V., Conti C., Roma E., Rao F., Sansone V. **(Milano)**

P.60 Expanded [CCTG]n repetitions are not associated with abnormal methylation at the CNBP locus in myotonic dystrophy type 2 (DM2) patients
Santoro M., Maiorca M., Fontana L., Centofanti F., Russo S., Massa R., Silvestri G., Novelli G., Botta A. **(Milano, Roma)**

P.61 Clinical variability in myotonic dystrophy type 1: a five-categories disease classification fits clinical but not brain complexity
Simoncini C., Baldanzi S., Ricci G., Cecchi P., Migaleddu G., Cosottini M., Siciliano G. **(Pisa)**

P.62 Evidence of mitochondrial dysfunction delays the diagnosis of myotonic dystrophy type 2
Valentino M.L., La Morgia C., Pellegrini C., Caporali L., Lodi R., Liguori R., Carelli V. **(Bologna)**

P.63 TP-PCR as a secondary analytical level in Myotonic Dystrophies diagnostic pathway
Lucchiari S., Conti B., Pagliarani S., Brusa R., Magri F., Govoni A., Peverelli L., Comi G.P. **(Milano)**

P.64 Cardiological assessment in a cohort of patients affected by congenital Myotonic Dystrophy type 1
Petillo R., D'Ambrosio P., Scutifero M., Orsini C., Palladino A., Politano L. **(Napoli)**

P.65 Mutational variety in patients with Myotonia Congenita from Campania Region
D'Ambrosio P., Petillo R., Scutifero M., De Luca C., Picillo E., Ergoli M., Passamano L., De Luca A., Politano L. **(Napoli, Roma)**

P.66 A novel mutation in KV1.1 channels in a patient with paroxysmal ataxia, myokymia, painful contractures and metabolic dysfunctions
Imbrici P., Altamura C., Gualandi F., Mangiatordi G.F., Neri M., De Maria G., Ferlini A., Padovani A., Sahbani D., D'Adamo M.C., Nicolotti O., Pessia M., Conte D., Filosto M., Desaphy J.F.
(Bari, Ferrara, Brescia, Msida, Perugia)

P.67 Periodic paralysis: an emergency department presentation
Agazzi E., Rottoli M.R. **(Bergamo)**

P2-2 DYSTROPHINOPATHY/LGMD/CONGENITAL MUSCULAR DYSTROPHIES

P.68 Hippo signaling pathway in muscular dystrophies
Aguennouz M., Polito F., Rodolico C., Vita G.L., Migliorato A., Oteri R., Ciranni A.M., Messina S., Toscano A., Di Giorgio R.M., Vita G. **(Messina)**

P.69 Circadian rhythm genes in Duchenne muscular dystrophy
Armaroli A., Osman H., Scotton C., Falzarano M.S., Rossi R., Capogrosso R.F., Cozzoli A., Camerino G.M., Schwartz E., De Luca A., Ferlini A. **(Ferrara, Bari, Rockville, London)**

P.70 LGMD2B with high dysferlin retention: two case reports
Barresi R., Hudson J., Hilton-Jones D., Petty R., Longman C., Farrugia M.E., Marini-Bettolo C.
(Newcastle upon Tyne, Oxford, Glasgow)

P.71 Plectin mutation without skin involvement as a possible cause of CMD
Berardinelli A., Rossi M., Ciscato P., Tironi R., Pichiecchio A., Cassandrini D., Santorelli F.M.
(Pavia, Milano, Pisa)

P.72 A rare case of myopathy with Pipestem capillaries in a female carrier of Becker muscular dystrophy
Cosentino G., Fierro B., Brighina F., Mirabella M., Rodolico C. **(Palermo, Roma, Messina)**

P.73 Effects of Long-Term Treatment with Eteplirsen on Cardiac Function: Left Ventricular Ejection Fraction in Eteplirsen-Treated Patients vs Disease Natural History
Duda P. W., Moody S., Colan S., Dworzak J., Mendell J.R. **(Cambridge, Durham, Boston, Columbus)**

P.74 Management of adult DMD patients: the experience of neuromuscular unit IRCCS e Medea
Gandossini S., Diella E., Scarpazza P., Marchi E., Russo A., Sala S., Lo Mauro A., Comi G.P., Aliverti A., D'Angelo M.G. **(Bosisio Parini, Vimercate, Milano)**

P.75 A case of limb-girdle muscular dystrophy type 2L mimicking Dermatomyositis
Gemelli C., Fiorillo C., Fabbri S., Cabona C., Zara F., Madia F., Mandich P., Grandis M. **(Genova)**

P.76 Custom micro-fluidic exome array to detect transcript mutations in undiagnosed patients with Collagen VI myopathies
Osman H., Rossi R., Falzarano M.S., Scotton C., Armaroli A., Selvatici R., Gualandi F., Ferlini A.
(Ferrara, Khartoum, London)

P.77 Muscle ultrasound elastography and MRI in preschool children with Duchenne muscular dystrophy: a pilot study
Pichiecchio A., Alessandrino F., Bortolotto C., Cerica A., Rosti C., Raciti M.V., Rossi M., Baranello G., Bastianello S., Berardinelli A., Calliada F. **(Pavia, Milano)**

P.78 Cognitive and psychiatric alterations in facioscapulohumeral muscular dystrophy: a case report
Pizzamiglio C., Solara V., Cantello R., Mazzini L. **(Novara)**

P.79 Distrofia facioscapolo omerale e syndrome 18p
Capet N., Renard D., Lagha N., Garibaldi M., Khau Van Kien P., Feasson L., Manel V., Cristofari G., Sacconi S. **(Nizza, Nimes, Lione, Saint Etienne)**

P.80 Respiratory pattern in FSHD patients as possible outcome measure
Scarlato M., Pozzi M., Velardo D., D'Angelo M.G., Pasanisi B., Maggi L., Previtali S.C., Tettamanti A.
(Milano, Bosisio Parini)

P.81 International-DMD (IDMD): a PTC therapeutics-supported diagnostic project to widely identify dystrophin mutations by NGS technologies
Selvatici R., Rossi R., Trabanelli C., Rimessi P., Fini S., Gualandi F., Ferlini A. **(Ferrara)**

P.82 Epidemiology of facioscapulohumeral muscular dystrophy in Abruzzo
Telese R., Tupler R., Di Muzio A. **(Chieti, Modena)**

P.83 Next-generation sequencing analysis for the diagnosis of Duchenne/Becker muscular dystrophies
Trabanelli C., Selvatici R., Rimessi P., Venturoli A., Fini S., Fabris M., Neri M., Gualandi F., Ferlini A.
(Ferrara)

P.84 Clinical and molecular consequences of EXON 78 deletion in DMD gene
Traverso M., Assereto S., Baratto S., Iacomino M., Pedemonte M., Diana M.C., Ferretti M., Bruno C., Zara F., Broda P., Minetti C., Gazzerro E.,Madia F., Fiorillo C. **(Genova)**

P.85 Multi-parametric characterization of highly fat infiltrated Limb Girdle muscular dystrophy patients: results of a multi-variate analysis
Velardo D., De Luca A., Bertoldo A., Froeling M., Gandossini S., Russo A., Magri F., Arrigoni F., D'Angelo M.G. **(Bosisio Parini, Padova, Utrecht, Milano)**

P.86 A diagnostic anoctamin-5 western blot
Vihola A., Luque H., Savarese M., Penttilä S., Lindfors M., Leturcq F., Eymard B., Tasca G., Brais B., Conte T., Charton K., Richard I., Udd B. **(Helsinki, Tampere, Paris, Roma, Montreal, Evry, Vaasa)**

P.87 Muscle magnetic resonance imaging as a prognostic biomarker in Becker muscular dystrophy
Bello L., Barp A., Caumo L., Campadello P., Semplicini C., Lazzarotto A., Zanato R., Stramare R., Pegoraro E. **(Messina, Roma)**

P.88 Effectiveness of treatment with ivabradine on clinical and instrumental endpoints in patients with Duchenne Muscular Dystrophy
Distefano M.G., Vita G.L., Sframeli M., Di Bella G., Pugliatti P., Recupero A., Barcellona C., La Rosa M., Nicocia G, Carerj S., Lunetta C., Messina S., Vita G. **(Messina)**

P.89 Cardiac involvement in a patient with congenital-muscular-dystrophy related to POMT2 gene mutation.
La Rosa M., Sframeli M., Distefano M.G., Barcellona C., Vita G.L., Nicocia G., Astrea G., D'Amico A., Bertini E., Santorelli F., Vita G., Lunetta C., Messina S. **(Messina, Pisa, Roma)**

P.90 Ten years (2006-2016) of molecular diagnosis in COLLAGEN-VI related myopathies: are intragenic coding SNPS of COL6A GENES modifyers of disease severity?
Rossi R., Trabanelli C., Venturoli A., Scotton C., Armaroli A., D'Amico A., Berardinelli A., Filosto M., Fiorillo C., Bruno C., Marrosu G., Pane M., Rodolico C., Mongini T., Moroni I., Baranello G., Santoro L., Pegoraro E., Politano L., Pini A., Fusco C., Merlini L., Morandi L., Bertini E., Messina S., Mercuri E., Fini S., Ferlini A., Gualandi F.
(Ferrara, Roma, Pavia, Brescia, Genova, Cagliari, Messina, Torino, Milano, Napoli, Padova, Caserta, Bologna, Reggio Emilia)

P.91 Congenital muscular dystrophy and epilepsy: a prospective observational study on 16 pediatric patients
Vitaliti G., Romano C., Sciuto C., Ruggieri M., Falsaperla R. **(Catania)**

P.92 Pivotal role of the clinical geneticist in diagnosing rare diseases. The index case of laminopathies
Passamano L., D'Ambrosio P., Petillo R., Della Pepa C., De Luca C., Torella A., Papa A.A., Palladino A., Novelli A., Nigro V., Politano L. **(Napoli, Roma)**

P2-3 SMA

P.93 Segmental body composition in young children with SMA type 2: correlation with motor function abilities
Baranello G., Arnoldi M.T., Zanin R., Masson R., Mastella C., De Amicis R., Battezzati A., Bertoli S. **(Milano)**

P.94 Fasting glucose in children with spinal muscular atrophy type I and II
Bertoli S., De Amicis R., Foppiani A., Leone A., Battezzati A. **(Milano)**

P.95 Scoliosis is an inescapable comorbidity in SMA type II. A single center experience
Catteruccia M., Colia G., Bonetti A.M., Carlesi A., Oggiano L., La Rosa G., Turturro F., Bertini E., D'Amico A. **(Roma)**

P.96 Longitudinal assessments in discordant twins with SMA
Palermo C., Pane M., Abiusi E., Lapenta L., De Sanctis R., Luigetti M., Ranalli D., Fiori S., Tiziano F., Mercuri E. **(Roma)**

P.97 Improved distal spinal muscular atrophy genetic diagnosis by targeted NGS sequencing
Moro F., Rubegni A., Lenzi S., Trovato R., Astrea G., Battini R., Battisti C., Bruno C., DiFabio R., Fiorillo C., Gallone S., Malandrini A., Mari F., Massa R., Pegoraro,E., Petrucci A., Pini A., Santorelli F.M.
(Roma, Siena, Genova, Torino, Firenze, Padova, Bologna)

P.98 Spinal muscular atrophy type 2 and 3: evaluation of autonomic nervous system function

Sframeli M.*, *Stancanelli C.*, *Vita G.L.*, *Terranova C.*, *Rizzo E.*, *Cavallaro F.*, *Lunetta C.*, *Vita G.*, *Messina S. **(Messina, Palermo)**

P.99 Improving SMARD1 long-term outcome and quality of life: the role of a multidisciplinary setting

Bonanno C.*, *Sframeli M.*, *Vita G.L.*, *Distefano M.G.*, *La Rosa M.*, *Barcellona C.*, *Profazio C.*, *Versaci A.*, *Mercurio L*, *Gitto E.*, *Romeo C.*, *Vita G.*, *Lunetta C.*, *Messina S. **(Messina)**